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Brief Clinical Report

Laryngeal Malformations in the Richieri-Costa and Pereira Form of Acrofacial Dysostosis

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We report on laryngeal malformations in 5 subjects, 4 females and 1 male, with the autosomal-recessive Richieri-Costa and Pereira form of acrofacial dysostosis. Characteristics of the voice are described.

KEY WORDS: acrofacial dysostosis, laryngeal malformations, voice disorders

INTRODUCTION

A new autosomal-recessive syndrome with short stature, Robin sequence, cleft mandible, pre- and postaxial hand anomalies, and clubfeet was reported in females [Richieri-Costa and Pereira, 1992] and in males [Richieri-Costa and Pereira, 1993]. In a recent review by Opitz et al. [1993], the syndrome here reported was classified as a "related" condition within the acrofacial dysostoses. Phoniatric examination of these patients demonstrated functional vocal disorders, and direct laryngoscopy, using a flexible nasolaryngoscope, demonstrated structural laryngeal anomalies. Here we report on these findings.

CLINICAL REPORTS

The 5 patients examined had the typical signs of the acrofacial dysostosis condition cited above: short stature, Robin sequence, cleft mandible, pre- and postaxial hand anomalies, and clubfeet. Patients 1, 2, and 5 were reported previously [Richieri-Costa and Pereira, 1992, 1993].

Patient 1

J.M.M.C., a 14-year-old boy, had, in addition to acrofacial dysostosis, a weak, slightly breathy, and harsh voice.

Laryngoscopy (Fig. 1) showed a short, round larynx, with a small epiglottis. The aryepiglottic folds were hypertrophic, making it impossible to identify the eminence of the arytenoids posteriorly. During emission of the vowel /i/, there was a medialization of the aryepiglottic folds which participate in phonation, and the vocal folds could not be seen.

Patient 2

V.L.V., a 7-year-old girl, had, in addition to acrofacial dysostosis, a weak, slightly breathy, and harsh voice. Laryngoscopy (Fig. 2) showed a short, round larynx and absence of the epiglottis. The aryepiglottic folds were hypertrophic, and it was not possible to identify the eminence of the arytenoids posteriorly. There was a fold situated above the level of the glottis in the posterior part of the larynx. During emission of the vowel /i/, there was a medialization of the aryepiglottic folds which participate in phonation, and the vocal folds could not be seen.

Patient 3

D.R.O., an 11-year-old girl (Fig. 3), also had a weak, mildly breathy, and slightly harsh voice.

Laryngoscopy (Fig. 4) showed a short, round larynx and a reduced epiglottis. The aryepiglottic folds were hypertrophic, and it was not possible to identify the eminence of the arytenoids posteriorly. There was a small fold situated above the level of the glottis in the posterior part of the larynx. During emission of the vowel /i/, there was a medialization of the aryepiglottic folds which participate in phonation, and it was only possible to see the median and anterior third of the vocal folds.

Patient 4

A.S.O., an 8-year-old girl (Fig. 5), also had a weak, mildly breathy, and slightly harsh voice. Laryngoscopy (Fig. 6) showed a short, round larynx and two small pieces of epiglottis at the base of the tongue. The aryepiglottic folds were hypertrophic, and it was impossible to identify the eminence of the arytenoids posteriorly. A fold situated above the level of the glottis

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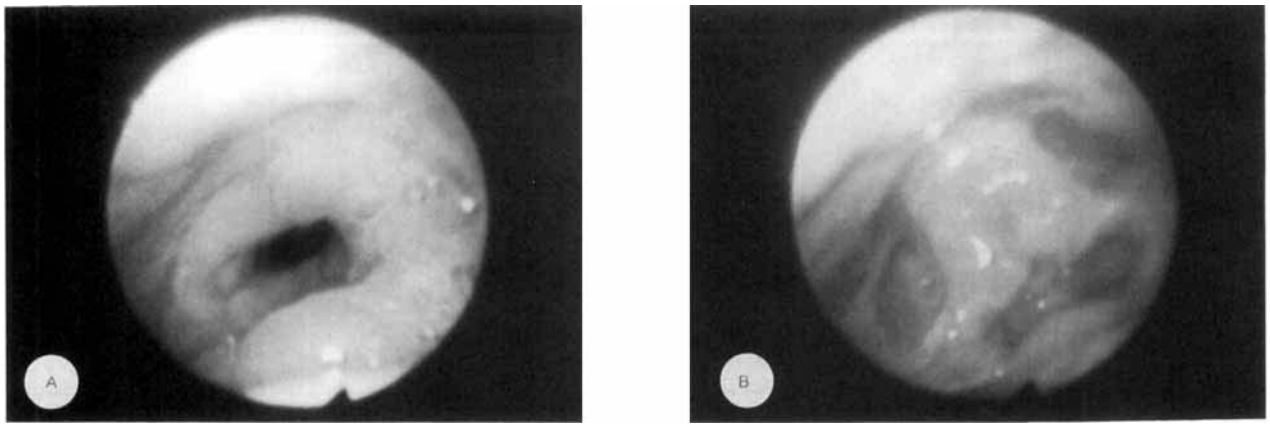

Fig. 1. Larynx of patient 1. **A**: Respiration. **B**: Emission of vowel /i/.

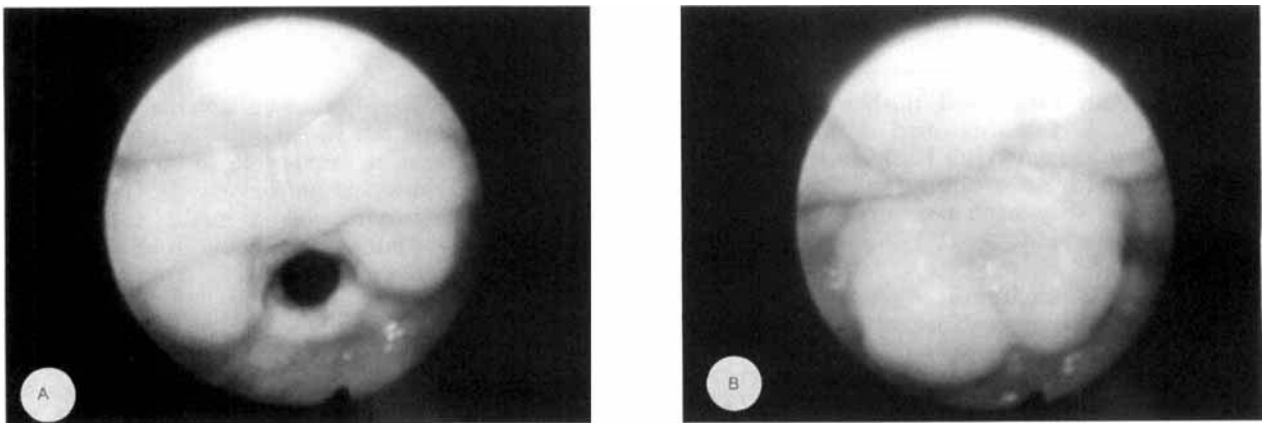

Fig. 2. Larynx of patient 2. **A**: Respiration. **B**: Emission of vowel /i/.

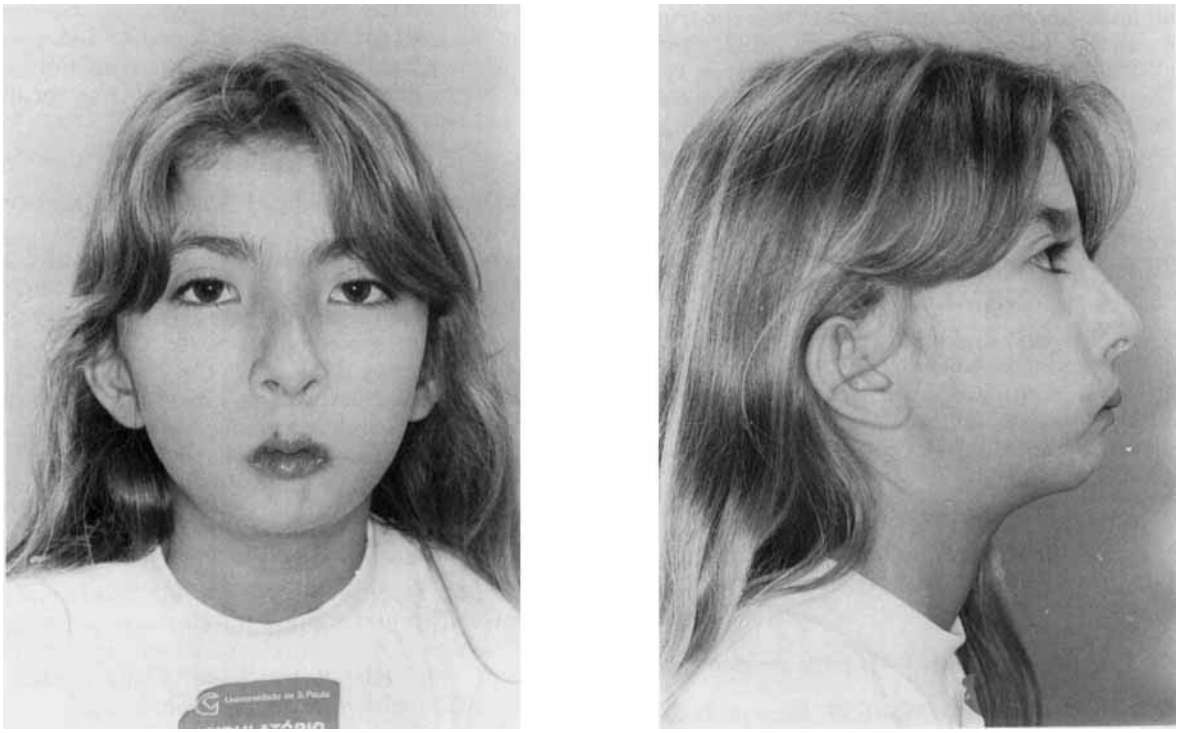

Fig. 3. Face of patient 3. Front and profile.

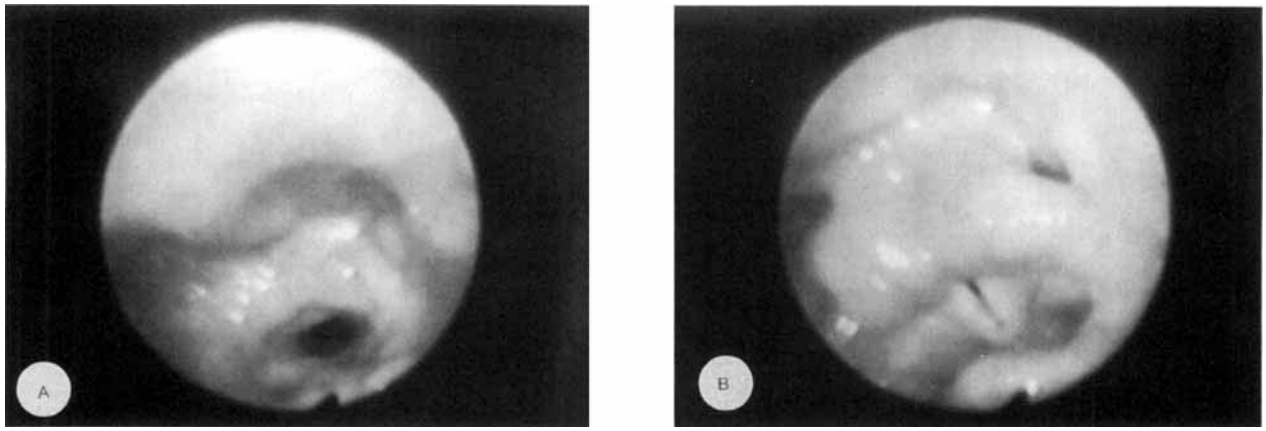

Fig. 4. Larynx of patient 3. **A**: Respiration. **B**: Emission of vowel /i/.

Fig. 5. Face of patient 4. Front and profile.

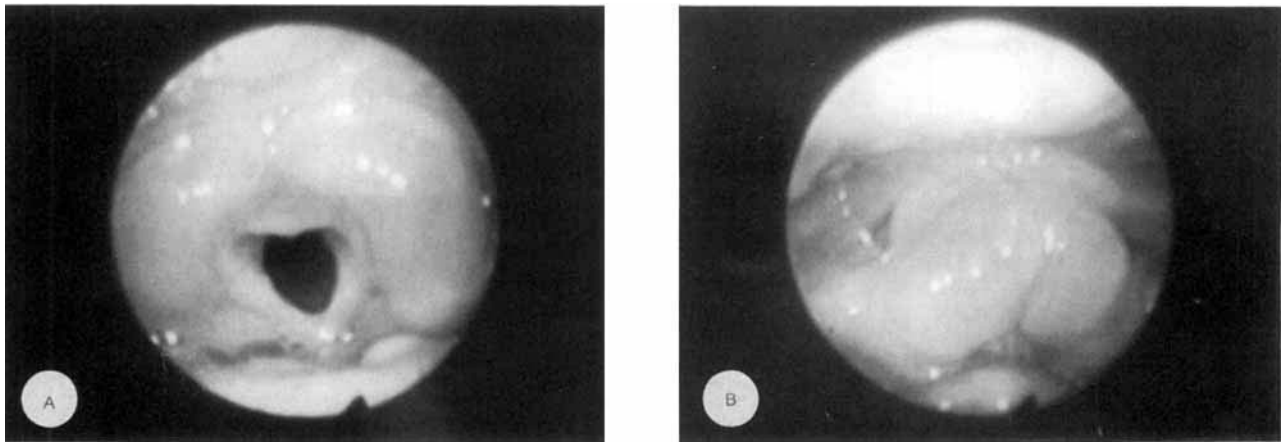

Fig. 6. Larynx of patient 4. **A:** Respiration. **B:** Emission of vowel /i/.

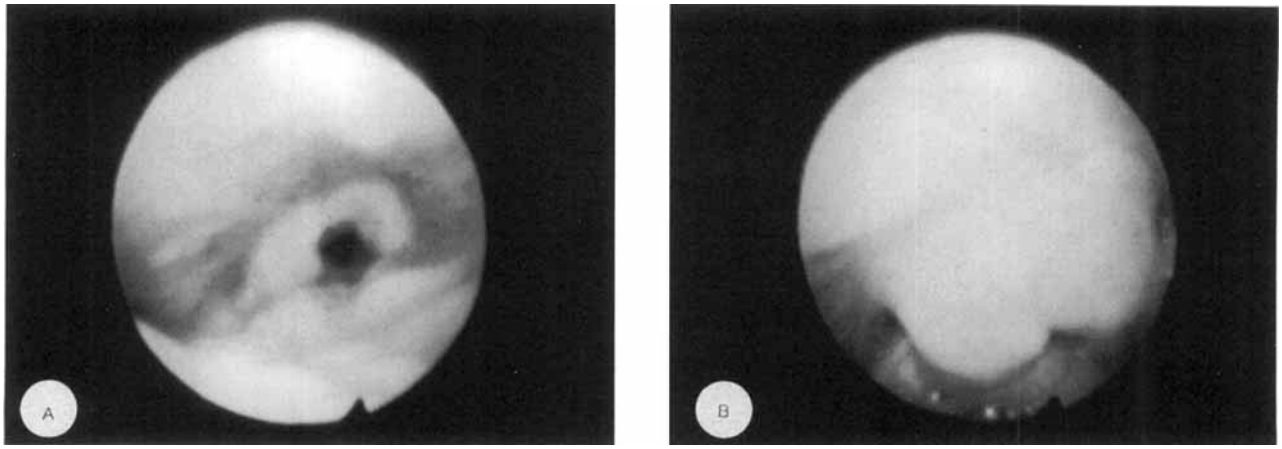

Fig. 7. Larynx of patient 5. **A:** Respiration. **B:** Emission of vowel /i/.

could be seen in the posterior part of the larynx. During emission of the vowel /i/, there was a medialization of the aryepiglottic folds which participate in phonation, and it was only possible to see part of the anterior third of the vocal folds.

Patient 5

N.O.S., a 9-year-old girl, also had a weak, slightly breathy, and harsh voice. Laryngoscopy (Fig. 7) showed a short, round larynx and absence of the epiglottis. The aryepiglottic folds were hypertrophic, and it was not possible to identify the eminence of the arytenoids posteriorly. There was a fold situated above the level of the glottis in the posterior part of the larynx. During emission of the vowel /i/, there was a medialization of the aryepiglottic folds which participate in phonation, making it impossible to see the vocal folds.

DISCUSSION

These findings indicate the presence of laryngeal malformations in this form of acrofacial dysostosis. All patients examined had the same laryngeal anomalies with variable severity: short, round larynx, absent or reduced epiglottis, hypertrophic aryepiglottic folds, and a fold in the posterior part of the larynx situated above the level of the glottis. During emission of sounds there was a medialization of the aryepiglottic folds, which probably also means a protection of the inferior airways while swallowing. These anatomic and functional abnormalities result in the vocal disorders characteristic of this autosomal-recessive syndrome.

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